Table 1. Mutations identified in the candidate gene in persons with Bloom's syndrome.

Person			Mutation					
I.D.[a]	Ancestry	Cell line	Position[b] (bp)	Alteration[c]	Zygosity at *BLM*[d]	Kind	Codon change	Predicted peptide[e]
97(AsOk)	Japanese	HG1926	631	3-bp del[f]	Homo	Nonsense	S→stop	185
112(NaSch)	German	HG2510	888	A→T	Hetero	Nonsense	K→stop	271
93(YoYa)	Japanese	HG1626	1610	1 bp ins	Homo	Frameshift[g]		515
139(ViKre)	American/European	HG2231	2089	A→G	Hetero	Missense	Q→R[h]	1417
15(MaRo)	Ashkenazi Jewish	HG1514	2281	6 bp del/ 7 bp ins	Homo	Frameshift[i]		739
42(RaFr)	Ashkenazi Jewish	HG2522	2281	6 bp del/ 7 bp ins	Homo	Frameshift[i]		739
107(MyAsa)	Ashkenazi Jewish	HG2654	2281	6 bp del/ 7 bp ins	Homo	Frameshift[i]		739
NR2(CrSpe)	Ashkenazi Jewish	HG2727	2281	6 bp del/ 7 bp ins	Homo	Frameshift[i]		739
92(VaBi)	Italian	HG1584	2596	T→C	Homo	Missense	I→T[j]	1417
113(DaDem)	Italian	HG1624	3238	G→C	Homo	Missense	C→S[k]	1417

[a] Bloom's Syndrome Registry designations. Three unrelated persons with BS were examined in whom mutations have yet to be detected: 61(DoHo), in HG2122; 30(MaKa), in HG1987; 140(DrKas), in HG1972.

[b] The nucleotide positions are as identified in the H1-5' sequence (Fig. 2).

[c] Del, deletion; ins, insertion.

[d] Homo, homozygous; hetero, heterozygous.

[e] Number of amino acids starting from the first in-frame ATG found in the H1-5' sequence (Fig. 2).

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Informal Ex. Amdt.

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* all notations by Examiner are